When reviewed at five months of age he had failed to grow satisfactorily. His weight was 3·5 kg (1·5 kg < 3rd centile), length 58 cm (3rd centile), and OFC 37·8 cm (2 cm < 3rd centile).

The following features were noted on examination: prominent epicanthic folds, long philtrum, high palate, micrognathia, a narrow upper lip, and low set, posteriorly rotated ears. He was having intermittent upper airway obstruction. A choanaogram showed bilateral choanal hypoplasia with stenosis and he required a tracheostomy to prevent further episodes of upper airways occlusion.

At review at two years he was still of short stature. His weight was 7·25 kg (2·5 < 3rd centile) and his OFC 44 cm (2 cm < 3rd centile). His development showed mild global delay. His facial appearance is shown in fig 2. He had short fingers with broad, flat nails. His chromosome analysis on two occasions has shown a normal 46,XY male karyotype. The parents have had no further children.

Discussion

The differential diagnosis of a child with congenital heart disease, growth failure, and mental retardation includes the effects to the fetus caused by maternal ingestion of hydantoins, valproate, and alcohol, Williams's syndrome,¹ the CHARGE association,² and the Smith-Lemli-Opitz (SLO) syndrome.³ However, the mother took no drugs or alcohol during the pregnancies and both Williams's syndrome and the CHARGE association are sporadic. SLO is an autosomal recessive condition, but the development of the two children reported here is better than that seen in SLO, the facial appearance is different, and the affected boy reported here did not have a genital anomaly.

We believe the two children reported here have the same syndrome. It is likely that the girl who died had mild choanal stenosis because she had significant problems in feeding and sucking in the first few months of life. The main features of this condition are, therefore, short stature, microcephaly, mild to moderate developmental delay, atrial and ventricular septal defects, choanal stenosis, and a distinctive facial appearance with marked epicanthic folds, a small nose, long philtrum, and narrow upper lip. The parents are not related. We propose an autosomal recessive mode of inheritance for this syndrome, though a submicroscopic chromosomal abnormality or germ line mosaicism for an autosomal dominant condition cannot be excluded at present.

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Friononal dysplasia, congenital heart defect, and short stature: a further observation

SUMMARY We present a mildly retarded boy with frontonasal dysplasia, valvar aortic stenosis, short stature, and small head circumference. In addition, mild genital anomalies and bilateral Sydney lines were present. Strikingly similar cases recently published by de Moor et al¹ suggest a defined clinical entity.

History

Prenatal. Low oestriol levels in third trimester, caesarean section at 35 weeks of gestation. Male newborn, small and light for dates.

Family. First child of healthy, non-consanguineous parents aged 33 years (mother) and 35 years (father). Healthy, one year old sister.

Clinical examination

At birth. Length 41 cm (3rd centile), weight 1810 g (10th centile), head circumference 29·5 cm (3rd centile). Hypertelorism with telecanthus, very broad nasal bridge, and broad, flat nasal tip. Small penis. At two years two months. Length 80 cm (−3 SD), weight 9·5 kg (3rd centile in relation to length), head circumference 45·5 cm (−2·6 SD for age, −2 SD in relation to length). Round face with small skull, hypertelorism with telecanthus (inner canthal distance 30 mm, 97th centile in relation to head circumference). Broad and flat nasal bridge with very broad and flat tip. Loud systolic murmur. Small penis, shawl scrotum, testes descended. Sydney lines bilaterally. Psychomotor development slightly retarded. Radiograph of the left hand showed markedly retarded skeletal maturation, corresponding to six months of age.

Medical history

Early infancy complicated by apnoeic episodes and frequent vomiting, improving gradually. Congenital heart defect suspected; later valvar aortic stenosis diagnosed.

Received for publication 15 November 1988.

Revised version accepted for publication 3 January 1989.

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FIG 1  Boy aged two years two months with facial features of frontonasal dysplasia.

by sonography; heart failure not present. Postnatal growth rate moderately diminished; later some catch up growth. Psychomotor and speech development slightly retarded.

Discussion

There is a striking similarity between this patient and those recently reported by de Moor et al. Their three unrelated patients also showed frontonasal dysplasia, congenital heart defect (CHD), short stature, and small head circumference. The main difference is the type of CHD; while the three published cases all had tetralogy of Fallot, our patient had valvular aortic stenosis. This may reflect clinical variability as is seen in several well established multiple congenital anomaly syndromes, for example, Noonan's syndrome or the velocardiofacial (Shprintzen) syndrome. Although cardiac anomalies have been documented in association with frontonasal dysplasia, a condition as described by de Moor et al probably represents a separate clinical entity.

Growth retardation apparently is part of this syndrome and cannot be explained by the CHD in the patients reported by de Moor et al or in our patient.

Other features, such as mild genital or dermatoglyphic anomalies in our patient, or brachycephaly, preauricular skin tags, clinodactyly, and cryptorchidism in the patients reported by de Moor et al may also belong to this syndrome or may be coincidental. The full range of variability of this syndrome will be learnt from more reports. In addition, long term prognosis is still unknown, as are the pathogenesis and aetiology. Though all cases so far published have been sporadic, a genetic aetiology cannot be excluded.

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